An 18-year-old woman presented with color and temperature changes in her hands, as well as intermittent tremor of the hands since the age of 15 years. She also reported involuntary right arm movements and difficulties with concentration. Physical examination of the eyes revealed bilateral Kayser–Fleischer rings (Panel A, arrows). Neurologic examination revealed dystonia of the right arm, a postural tremor of her arms and legs, mild dysphagia and dysarthria, and bradykinesia. Laboratory tests revealed elevated serum levels of alanine aminotransferase, aspartate aminotransferase, and γ-glutamyltransferase, as well as low serum levels of ceruloplasmin (0.02 g per liter; reference range, 0.2 to 0.5) and copper (4.1 µmol per liter; reference range, 11 to 22); the urinary copper excretion was elevated, at 12.8 µmol per 24 hours (reference range, 0 to 1). Magnetic resonance imaging of the patient’s brain revealed widespread signal change, gliosis, and atrophy in the basal ganglia, thalami, and brain stem. A diagnosis of Wilson’s disease was made and confirmed on genetic testing by a result of a compound heterozygous mutation in the gene ATP7B. Approximately 5 years after the initiation of iron-chelating treatment, the Kayser–Fleischer rings had resolved almost completely (Panel B), and there was stabilization in neurologic status and MRI findings.

Kayser–Fleischer Rings in Wilson’s Disease

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